- **GRAF, Justin Troy**

  *BAppSci (Hons) QUT*

  **Thesis Title:**

  Membrane Associated Transporter Protein Gene (*SLC45A2*) and the Genetic Basis of Normal Human Pigmentation Variation.

  **Supervisors:**

  Dr Ian Hughes (Principal)
  Dr Joanne Voisey (Associate)

  **Citation:**

  *This research demonstrated that normal variation in hair, skin, and eye colour, both within and between human populations, is determined by small, common differences in the DNA. Specifically, it revealed that DNA differences in coding and control regions of SLC45A2 are associated with normal pigmentation variation within a Caucasian population and then investigated possible biological mechanisms responsible for this.* Dr Graf chose to study *SLC45A2*, as it was known that certain mutations in this gene cause a form of albinism. Common polymorphisms known not to cause albinism were identified and investigated. Four of these, in both the promoter and coding regions, were shown to be associated with pigmentation variation in a Caucasian population. Two promoter polymorphisms were shown to affect the transcription of *SLC45A2* and, interestingly, the expression of other pigmentation genes. This work has implications for forensics, anthropology, treatment of pigmentation disorders, and skin cancer susceptibility.